Case Report

Spine duplication or split notochord syndrome – case report and literature review

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Context: Spine duplication is a rare condition, with various extents and severe additional anomalies. The goal of this study was to describe a unique case of a boy with split notochord syndrome who was followed up from birth until maturity.

Findings: Physical examination at birth showed defects of the abdominal wall and cloacal exstrophy with visible urether outlets. A transposed anus was present in the perineal region. Split bony elements of the spine with nonpalpable sacral bone were noted. A soft, skin-covered lump, with the consistency of a lipoma, was present in the sacral area. There was asymmetry of the lower limbs: the left was hypoplastic, with a deformed foot and hip. Computed tomography revealed a normal shape of the Th12 and L1 vertebrae, whereas the L2 was split. Downward from L3, there were two vertebrae at each level, with two spinal canals. The spinal cord divided into two "semicords" at the level of L1. Neurologic status and the shape of the spine remained unchanged during puberty. The last follow-up was performed at the age of 18 years. He managed to walk independently in prosthesis with visible limping.

Conclusion: Spine deformities are always suspected in neonates with lipoma in the sacral region, which may sometimes be serious. Walking ability and quality of life depend on neurologic deficits; even with long duplication and double sacrum, walking can be a feasible option.

Keywords: Spine duplication, Split notochord syndrome, Split cord malformation, Congenital spine defect

Introduction

Spine duplication is a rare condition, with various extents of duplication and various additional anomalies. Two parallel names coexist in the literature: "split notochord syndrome" and "spinal duplication syndrome."[1–3] At the end of the nineteenth-century, Rembe described dorsal enteric fistula for the first time.[1] Since then, literature on a combination of congenital defects, including some intestinal (enteric cysts, fistulas), vertebral (duplication), and central nervous anomalies (myelomeningocele), has been published, usually as case reports of single patients.[4–37] The name "split notochord syndrome" was proposed by Bentley and Smith[6] for lesions caused by partial duplication or separation of the notochord. Dominguez *et al.*[20] proposed the name "caudal duplication syndrome," which resulted from the deformation of the caudal cell mass and hindgut.[20] In some cases, the problems with spinal duplication overshadowed those associated with the gastrointestinal tract, and for these cases, the name "spine duplication syndrome" was used. According to Dias and Pang,[38] it was thought to be an extreme form of split cord malformation.

The goal of this study was to describe a unique case – the observation and treatment of a boy with split notochord syndrome followed from birth until his skeletal maturity. To our knowledge, there is no such observation or follow-up period recorded in the literature.

Materials

We describe the case of a male infant, born on time by cesarean section (due to breech position). Congenital defects of the spine were associated with anomalies of the gastrointestinal and urogenital tracts.

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Methods

Retrospective analysis of medical data.

Results

A male neonate weighing 2800 g was delivered at term by caesarian section. Apgar score was 1 at 1 min. His parents were unrelated, and the maternal history was uneventful. Physical examination showed defects of the abdominal wall in the underbelly, cloacal exstrophy with visible urether outlets. The penis and the scrotum were transposed to the right. A narrow fistula or transposed anus, with stool passing through it, was present in the perineal region. Further investigation revealed a normal left kidney and ectopis right . At the back, split bony elements of the spine with nonpalpable sacral bone were noted. A soft, skin-covered lump, with the consistency of a lipoma, was present in the sacral area. There was asymmetry of the lower limbs: the right lower limb had a normal shape and range of motion of the joints, whereas the left lower limb was hypoplastic, with a deformed foot and limited active and passive motions (flaccid paresis from the level of L4). Beginning at the first month of life, the patient underwent several surgeries because of urogenital and gastrointestinal defects.

Computed tomography performed in the second month of life revealed a normal shape of Th12 and L1 vertebrae, whereas the L2 was split. Downward from L3, there were two vertebrae at each level, with two spinal canals. Magnetic resonance imaging (MRI) done in the second year of life showed that the spinal cord was additionally divided into two "semicords" at the level of L1 vertebrae; in the lump, there were some neural structures with fibrous tissue and cerebrospinal liquid (Fig. 1). This finding was suspected to be a remnant of embryonic tissues.

A lipomyelocele-creating tumor/lump in the sacral area was resected by neurosurgeons at the age of 2 years. Rehabilitation of the patient was started as soon as possible, with standing and walking in prosthesis (due to the hypoplastic left leg). The first years of our patient were described by Radlo *et al.* in 1998.[37] During the first decade of life, the patient's primary problems were decubitus ulcers on the sacral and ischial areas and on the buttocks. The patient underwent several orthopedic interventions due to left hip subluxation and left clubfoot: foot correction–soft tissue release (at 3 years), proximal femoral osteotomy (at 7 years), and acetabuloplasty of the left hip (at 10 and 15 years). Hardware removal from the femur with simultaneous resection of exostosis and knee capsulotomy was performed when he was 17 years old.

The patient was examined regularly until skeletal maturity (by orthopedists and neurosurgeons). His neurologic status and shape of the spine were unchanged during puberty. Repeated spine X-ray and MRI showed consistently split posterior elements from Th11 and spine duplication, with two semicords, each in their own thecal sac, with a meningocele on the left (up to subcutaneous tissue) (Fig. 2). The last follow up examination was performed at the age of 18. His left leg was shorter, hypoplastic, and weak. He managed to walk independently in prosthesis with visible limping due to the lack of pelvic stability.

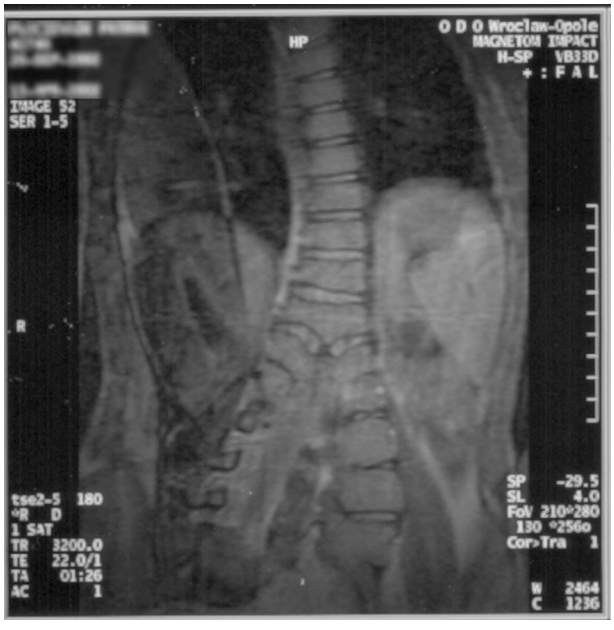

Figure 1 Magnetic resonance imaging of the spine duplication from Th11 downward.

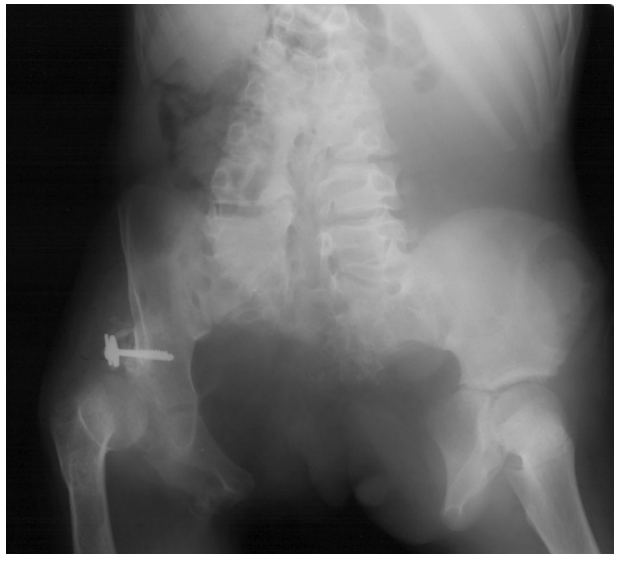

Figure 2 X-ray of the spine and pelvis at the age of 18 years.

Discussion

For years, pediatricians, surgeons, and orthopedists have described cases called either "split notochord syndrome" or "duplication of the spine," depending on the most visible defects in the child. In literature, more than 40 cases have been already mentioned.[4–37] The syndrome comprises a complete spina bifida (both anterior and posterior) with a dorsal opening of the hindgut.[22] In 1960, Bentley and Smith first underlined that "abnormal splitting of the notochord could cause a wide variety of malformations" involving the vertebral bodies, spinal cord, and enteric viscera.[6,18] When only spine defects are present, diagnosis could be delayed.[21,23,26] With enteric anomalies, the diagnosis is established just after birth, and the survival of these children depends on the severity of the visceral anomalies and fistulas.[14,18,25] In those cases, various patient-tailored surgical procedures are necessary to save the life and to enable further development, as seen in our case.[36,39] Our patient, despite surgeries due to visceral defects, underwent several orthopedic procedures aimed at improving independent walking.

Taking the spine into account, duplication can vary from splitting of only the sacrum and coccyx to duplication of the entire lumbar spine. Owing to the origin of the anomaly as a neural tube defect, it is usually related to myelomeningocele, which are semicords tethered to a filum lipoma.[3,23,26] Lumbar spine duplication may be classified as an extreme variant of split cord malformation with wide splitting of the neural tube and vertebrae.[26,38] The sacrum may be duplicated or reunited. Scoliosis and other spinal deformities may be apparent and a "lump" in the lower lumbar area due to lipoma may be present, suggesting intraspinal anomalies. Patients with spine duplication have various neurologic deficits. Considering our case and the literature, we observed two "principles" that would describe the prognosis. The first is that more visceral anomalies lead to more severe defects and poorer neurologic status.[4–37] The second principle concerns the sacral bone. For longer duplications, a reunited sacrum usually means no or minor neurologic deficits.[17,23,26] A patient with duplicated sacrum may have a normal neurologic status and walking ability if the split occurs only in the sacrum and the coccyx.[33] These principles may be helpful in establishing a prognosis in newborns in terms of their future life and development.

Spine duplication is not a typical split cord malformation, and most orthopedists do not perform any surgery, unlike in typical spilt cord cases.[2,21,23,26,35,38] The neurosurgical excision of the lipomyelocele in our patient was performed to enable rehabilitation and standing-walking in prosthesis on the hypoplastic limb. In previous studies, most authors presented their patients at one specific period of their life or the follow-up period did not exceed 5 years,[1,2,31,32,34–36] but our case report is unique because it shows the growth and development of the patient from birth to adulthood. A duplicated lumbar spine with two sacral bones indicates a bad prognosis in terms of walking ability. However, our patient's case is exceptional, as neurologic deficits were limited to one leg. We know now that the past decision of neurosurgical treatment was good, as the patient started to walk independently and the growth spurt did not worsen the neurologic status.

The limitation of the study is that it is a report of only one case – as are most papers published on this topic. With the existence of many types of spine duplication and various visceral anomalies, it is very difficult to draw any conclusions even on the basis of literature review. We would recommend further research with data collected from most authors of published case reports on this subject.

In conclusion, if spine duplication is seen in the prenatal period, the mother should be referred to a tertiary hospital due to the possible presence of visceral anomalies that can threaten the life of the fetus. Spine deformities are always suspected in cases with lipoma in the sacral region, which is not usually of utmost importance in the neonatal period. Walking ability and quality of life depend on neurologic deficits; even with long duplication and double sacrum, walking can be a feasible option.

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